WEST

End of Result Set

Generate Collection Print

L30: Entry 2 of 2 File: DWPI Feb 19, 1999

DERWENT-ACC-NO: 2000-098181
DERWENT-WEEK: 200009

TITLE: Identifying individuals at risk of developing spinocerebellar ataxia type 7 by analyzing trinucleotide repeat regions of spinocerebellar ataxia type 7 gene

INVENTOR: KOOB, M D; RANUM, L P ; BENZOW, K A ; MOSELEY-ALLDREDGE, M L ; RANUM, L P W

PATENT-ASSIGNEE:

ASSIGNEE	CODE
UNIV MINNESOTA	MINU

PRIORITY-DATA: 1997US-056170P (August 19, 1997), 1998US-0135994 (August 18, 1998)

PATENT-FAMILY:

PUB-NO	PUB-DATE	LANGUAGE	PAGES	MAIN-IPC
CA 2245310 A	February 19, 1999		064	C12N015/12
JP 11206393 A	August 3, 1999		091	C12N015/09
US 6280938 B1	August 28, 2001		000	C12Q001/68

APPLICATION-DATA:

PUB-NO	APPL-DATE	APPL-NO	DESCRIPTOR
CA 2245310A	August 19, 1998	1998CA-2245310	
JP 11206393A	August 19, 1998	1998JP-0294732	
US 6280938B1	August 19, 1997	1997US-056170P	Provisional
US 6280938B1	August 18, 1998	1998US-0135994	

INT-CL (IPC): C07 H 21/04; C07 K 14/47; C07 K 16/18; C12 N 15/09; C12 N 15/12; C12 P 21/02; C12 Q 1/68; G01 N 33/53; G01 N 33/566; G01 N 33/68

ABSTRACTED-PUB-NO: CA 2245310A
BASIC-ABSTRACT:

NOVELTY - A new method for identifying individuals at risk for developing spinocerebellar ataxia type 7 (SCA7) comprises analyzing the CAG repeat region of a SCA7 gene to detect CAG repeats, where individuals at risk have at least 30 CAG repeats and those not at risk have less than 19 CAG repeats.

DETAILED DESCRIPTION - INDEPENDENT CLAIMS are also included for the following:

(1) A kit for detecting whether or not an individual is at risk for developing SCA7 comprising selected from the 129 basepair (bp) (N1) and 192 bp (N2) regions of the 477 bp sequence (I) given in the specification;

(2) A method of detecting the presence of a DNA molecule located within an affected

allele of the SCA7 gene comprises:

(a) treating separate complementary strands of a DNA molecule containing a CAG repeat region of the SCA7 gene with a molar excess of two oligonucleotide primers;

(b) extending the primers to form complementary primer extension products which act as templates for synthesizing the desired molecule containing the CAG repeat region;

(c) detecting the molecule amplified; and

(d) analyzing the amplified DNA molecule for a CAG repeat region characteristic of the SCA7 disorder;

(3) A method of detecting the presence of a DNA molecule containing a CAG repeat region of the SCA7 gene comprises:

(a) digesting genomic DNA with restriction endonucleases to obtain DNA fragments;

(b) probing the fragments under hybridizing conditions with a detectably labeled gene probe, which hybridizes to a nucleic acid containing a CAG repeat region of an isolated SCA7 gene having at least 11 nucleotides;

(c) detecting the hybridized DNA fragments; and

(d) analyzing the DNA fragments for a CAG repeat region characteristic of the normal or affected forms of the SCA7 gene;

(4) A nucleic acid molecule containing a CAG repeat region of an isolated SCA7 gene, where the gene is located within the short arm of chromosome 3;

(5) An isolated DNA fragment comprising bases 1-128 of (I) and optionally further comprising a CAG repeat region, or bases 286-476 of (I);

(6) An isolated DNA fragment comprising bases 922-1002 of (II) and optionally further comprising a CAG repeat region, or bases 1033-1864 of (II);

(7) An isolated DNA fragment comprising bases 1-128 of (I) or bases 922-1002 of (II) in a vector;

(8) A polypeptide encoded by (I) or (II);

(9) An oligonucleotide comprising at least 15 nucleotides from N1, N2, N3 or N4;

(10) An isolated oligonucleotide that hybridizes to a nucleic acid molecule containing a CAG repeat region of an isolated SCA7 gene, where the oligonucleotide has at least 11 nucleotides;

(11) An isolated recombinant vector comprising (I) or (II) operatively linked to a heterologous vector sequence;

(12) An isolated nucleic acid fragment encoding a polypeptide for SCA7 comprising the 27 amino acid sequence of the 129 amino acid residue sequence given in the specification, followed by a polyglutamine repeat region;

(13) Cells containing the vector of (11);

(14) A protein encoded by the SCA7 gene having a glutamine repeat region;

(15) An antibody to a protein encoded by DNA containing a CAG repeat region of the SCA7 gene;

(16) A method for detecting the SCA7 disorder comprises:

(a) contacting an antibody to a SCA7 gene with a biological sample containing

antigenic protein to form an antibody-antigen complex;

(b) isolating the antibody-antigen complex; and

(c) sequencing the antigen portion of the antibody-antigen complex using amino acid sequencing techniques;

(17) A method for identifying expanded repeats from genomic DNA comprises:

(a) fractionating a population of DNA fragments and detecting the fraction that contains the expanded repeat;

(b) cloning the DNA fragments contained in the fraction of DNA that contains an expanded repeat; and

(c) identifying the clones that contain the expanded repeat;

(18) A method for identifying expanded trinucleotide repeats from genomic DNA comprising performing repeat expansion detection (RED) analysis on a sample of genomic DNA where the rate of temperature change from the denaturation temperature is decreased and where the ligation buffer contains formamide.

USE - The method is useful for identifying individuals at risk of developing SCA7 and also those at risk of developing SCA1, 2, 3 or 6.

ADVANTAGE - The use of genomic DNA in the repeat expansion detection (RED) analysis allows isolation of any potential trinucleotide repeat expansion regardless of the expression pattern. Utilization of different oligonucleotides in the RED assay allows any of the possible trinucleotide repeats to be detected, and the cycled nature of the reaction makes it extremely sensitive.

DESCRIPTION OF DRAWING(S) - The drawing shows the kindreds, diagnosed with autosomal dominant ataxia with retinopathy, which were used in PCR analysis of the spinocerebellar ataxia type 7 (SCA7) alleles. The estimated age of onset is in parentheses and number of CAG repeats is indicated numerically in each kindred.
ABSTRACTED-PUB-NO:

US 6280938B
EQUIVALENT-ABSTRACTS:

NOVELTY - A new method for identifying individuals at risk for developing spinocerebellar ataxia type 7 (SCA7) comprises analyzing the CAG repeat region of a SCA7 gene to detect CAG repeats, where individuals at risk have at least 30 CAG repeats and those not at risk have less than 19 CAG repeats.

DETAILED DESCRIPTION - INDEPENDENT CLAIMS are also included for the following:

(1) A kit for detecting whether or not an individual is at risk for developing SCA7 comprising selected from the 129 basepair (bp) (N1) and 192 bp (N2) regions of the 477 bp sequence (I) given in the specification;

(2) A method of detecting the presence of a DNA molecule located within an affected allele of the SCA7 gene comprises:

(a) treating separate complementary strands of a DNA molecule containing a CAG repeat region of the SCA7 gene with a molar excess of two oligonucleotide primers;

(b) extending the primers to form complementary primer extension products which act as templates for synthesizing the desired molecule containing the CAG repeat region;

(c) detecting the molecule amplified; and

(d) analyzing the amplified DNA molecule for a CAG repeat region characteristic of the SCA7 disorder;

(3) A method of detecting the presence of a DNA molecule containing a CAG repeat region of the SCA7 gene comprises:

(a) digesting genomic DNA with restriction endonucleases to obtain DNA fragments;

(b) probing the fragments under hybridizing conditions with a detectably labeled gene probe, which hybridizes to a nucleic acid containing a CAG repeat region of an isolated SCA7 gene having at least 11 nucleotides;

(c) detecting the hybridized DNA fragments; and

(d) analyzing the DNA fragments for a CAG repeat region characteristic of the normal or affected forms of the SCA7 gene;

(4) A nucleic acid molecule containing a CAG repeat region of an isolated SCA7 gene, where the gene is located within the short arm of chromosome 3;

(5) An isolated DNA fragment comprising bases 1-128 of (I) and optionally further comprising a CAG repeat region, or bases 286-476 of (I);

(6) An isolated DNA fragment comprising bases 922-1002 of (II) and optionally further comprising a CAG repeat region, or bases 1033-1864 of (II);

(7) An isolated DNA fragment comprising bases 1-128 of (I) or bases 922-1002 of (II) in a vector;

(8) A polypeptide encoded by (I) or (II);

(9) An oligonucleotide comprising at least 15 nucleotides from N1, N2, N3 or N4;

(10) An isolated oligonucleotide that hybridizes to a nucleic acid molecule containing a CAG repeat region of an isolated SCA7 gene, where the oligonucleotide has at least 11 nucleotides;

(11) An isolated recombinant vector comprising (I) or (II) operatively linked to a heterologous vector sequence;

(12) An isolated nucleic acid fragment encoding a polypeptide for SCA7 comprising the 27 amino acid sequence of the 129 amino acid residue sequence given in the specification, followed by a polyglutamine repeat region;

(13) Cells containing the vector of (11);

(14) A protein encoded by the SCA7 gene having a glutamine repeat region;

(15) An antibody to a protein encoded by DNA containing a CAG repeat region of the SCA7 gene;

(16) A method for detecting the SCA7 disorder comprises:

(a) contacting an antibody to a SCA7 gene with a biological sample containing antigenic protein to form an antibody-antigen complex;

(b) isolating the antibody-antigen complex; and

(c) sequencing the antigen portion of the antibody-antigen complex using amino acid sequencing techniques;

(17) A method for identifying expanded repeats from genomic DNA comprises:

(a) fractionating a population of DNA fragments and detecting the fraction that contains the expanded repeat;

(b) cloning the DNA fragments contained in the fraction of DNA that contains an

expanded repeat; and

(c) identifying the clones that contain the expanded repeat;

(18) A method for identifying expanded trinucleotide repeats from genomic DNA comprising performing repeat expansion detection (RED) analysis on a sample of genomic DNA where the rate of temperature change from the denaturation temperature is decreased and where the ligation buffer contains formamide.

USE - The method is useful for identifying individuals at risk of developing SCA7 and also those at risk of developing SCA1, 2, 3 or 6.

ADVANTAGE - The use of genomic DNA in the repeat expansion detection (RED) analysis allows isolation of any potential trinucleotide repeat expansion regardless of the expression pattern. Utilization of different oligonucleotides in the RED assay allows any of the possible trinucleotide repeats to be detected, and the cycled nature of the reaction makes it extremely sensitive.

DESCRIPTION OF DRAWING(S) - The drawing shows the kindreds, diagnosed with autosomal dominant ataxia with retinopathy, which were used in PCR analysis of the spinocerebellar ataxia type 7 (SCA7) alleles. The estimated age of onset is in parentheses and number of CAG repeats is indicated numerically in each kindred.

CHOSEN-DRAWING: Dwg.8/8

TITLE-TERMS: IDENTIFY INDIVIDUAL RISK DEVELOP TYPE REPEAT REGION TYPE GENE

DERWENT-CLASS: B04 D16 S03

CPI-CODES: B04-C01G; B04-E03F; B04-E05; B04-E08; B04-F0100E; B04-F1100E; B04-G01; B04-L05A; B04-N02A; B11-C07A; B11-C08D1; B11-C08E3; B11-C08E5; B12-K04A5; B12-K04F; D05-H09; D05-H11; D05-H12A; D05-H12D1; D05-H12E; D05-H14; D05-H17A6; D05-H18B;

EPI-CODES: S03-E14H; S03-E14H4;

CHEMICAL-CODES:

Chemical Indexing M1 *01*
Fragmentation Code
M423 M424 M710 M740 M905 N102 N161 Q233
Specfic Compounds
A00NSN

Chemical Indexing M1 *02*
Fragmentation Code
M423 M424 M710 M740 M905 N102 N161 Q233
Specfic Compounds
A00GTN

Chemical Indexing M1 *03*
Fragmentation Code
M423 M424 M710 M740 M905 N102 N161 Q233
Specfic Compounds
A00H3N

Chemical Indexing M1 *04*
Fragmentation Code
M423 M424 M740 M781 M905 N102 N134 N161 P831 Q233
Q505
Specfic Compounds
A00NSK A00NSD

Chemical Indexing M1 *05*
Fragmentation Code

M423 M424 M710 M740 M781 M905 N102 N134 N161 P831
Q233 Q505
Specfic Compounds
A013ID A013IN

Chemical Indexing M1 *06*
Fragmentation Code
M423 M424 M740 M750 M905 N102 N134 N161 P831 Q233
Specfic Compounds
A00NSK A00NSA

Chemical Indexing M1 *07*
Fragmentation Code
M423 M424 M710 M740 M781 M905 N102 N161 P831 Q233
Q505
Specfic Compounds
A00C8D A00C8N

Chemical Indexing M1 *08*
Fragmentation Code
M423 M424 M740 M750 M905 N102 N161 P831 Q233
Specfic Compounds
A00H3K A00H3A

Chemical Indexing M6 *09*
Fragmentation Code
M905 P831 Q233 Q505 R515 R521 R613 R621 R627 R631
R639

SECONDARY-ACC-NO:
CPI Secondary Accession Numbers: C2000-028607
Non-CPI Secondary Accession Numbers: N2000-075852